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Facultat d'Òptica i Optometria de Terrassa

GRAU EN ÒPTICA I OPTOMETRÍA

TREBALL FINAL DE GRAU

VISUAL AFFECTATION IN PATIENTS WITH GLYCOGEN STORAGE DISEASE (GLYCOGENOSIS)

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Terrassa, 15 de Gener 2016

GRAU EN OPTICA I OPTOMETRIA

La Sra. Montsrrrat Augé Serra, com a tutor/a del treball

CERTIFIQUEN

Que la Sra. Sònia Travé Huarte ha realitzat sota la seva supervisió el treball **Visual affectation in patients with glycogen storage disease (glycogenosis)** que es recull en aquesta memòria per optar al títol de grau en Òptica i Optometria.

I per a què consti, signo/em aquest certificat.

Sra. Montserrat Augé Serra
Directora del treball

Terrassa, 15 de Gener de 2016

GRAU EN OPTICA I OPTOMETRIA

VISUAL AFFECTATION IN PATIENTS WITH GLYCOGEN STORAGE DISEASE (GLYCOGENOSIS).

RESUM

En aquest treball hem volgut abarcar les afectacions d'una malaltia rara i minoritària a nivell ocular. Basant-nos en la poca recerca que hi havia en aquest tema i la gran afectació que aquesta pot tenir a nivell visual. Aquesta malaltia amb aquestes característiques no té el mateix nivell d'investigació que una malaltia com la Sida o la Malària ja que a nivell mundial és considerada com a malaltia rara, pel baix percentatge d'afectats. Poques subvencions són donades per la seva recerca.

Fent coincidir l'assignatura d'Optometria Geriàtrica i Infantil, on es tractaven els defectes binoculars i els estrabismes, (tant a nivell motor com acomodatiu), amb el 5è Congres Internacional de Glucogenosis, unint forces, dues estudiants de la FOOT i una professora Montserrat Augé i Serra, vàrem ser presents en el congres on vam tenir la sort de poder dur a terme exàmens visuals en pacients afectats amb la malaltia esmentada, tenint així gran varietat de pacients amb diferents subtipus de Glucogenosis.

Durant els dies 5,6,7 de juny de 2014, a Bellaterra va tenir lloc el 5è Congres de Glucogenosis a nivell Internacional, on apart d'especialistes nutricionals i mèdics, també hi havien doctors especialitzats en fetge i muscles. A cada assistent es donava una guia de les xerrades i dos llibres sobre l'afectació de glucogenosis a Espanya i sobre les noves teràpies pal·liatives, o no, trobades gràcies a la recerca, a part de certs productes alimentaris de prova per introduir a les dietes dels pacients.

GRAU EN OPTICA I OPTOMETRIA

VISUAL AFFECTATION IN PATIENTS WITH GLYCOGEN STORAGE DISEASE (GLYCOGENOSIS).

RESUMEN

En este trabajo queremos englobar las afectaciones visuales de una enfermedad rara/minoritária. Basandonos en la poca información que habia en este campo y la grande afectación que esta puede tener a nivel visual. Ésta enfermedad con éstas características no tiene el mismo nivel de investigación que una Sida o una Malária, ya que a nivel mundial es considerada enfermedad rara i/o minoritária, por el bajo porcentaje de afectados. Pocas subvenciones son dadas para su investigación.

Haciendo coincidir la asignatura de Optometría Geriátrica y Infantil, donde se trataban defectos binoculares y estrabismos (tanto a nivel motor como a nivel acomodativo), con el 5º Congreso Internacional de Glucogenosis, uniendo fuerzas, dos estudiantes de la FOOT I una profesora, Montserrat Augé i Serra, estuvieron presentes en el congreso donde tuvieron la suerte de poder llevar a cabo una serie de exámenes optométricos a pacientes afectados con la enfermedad susodicha teniendo así una gran muestra de pacientes con diferentes subtipos de Glucogenosis.

Durante los días 5,6,7 de Junio de 2014, en Bellaterra, tuvo lugar el 5º Congreso Internacional de Glucogenosis, donde aparte de nutricionistas y médicos, también habían doctores especializados en hígado y músculos. A cada asistente se le daba una guía de las conferencias y dos libros sobre la afectación de Glucogenosis en España y las nuevas terapias paliativas o no, descubiertas gracias a la investigación científica, aparte de ciertos productos alimenticios de prueba para introducir en las dietas de los pacientes afectados con Glucogenosis.

GRAU EN OPTICA I OPTOMETRIA

VISUAL AFFECTATION IN PATIENTS WITH GLYCOGEN STORAGE DISEASE (GLYCOGENOSIS).

SUMMARY

In this project we wanted to comprise the visual affectations of a rare/minority disease. Based on the little research there is in this field and the large effect that this may actually have in the visual sistem. We have to take into account that a disease like this one does not have the same level of research as AIDS or Malaria, due to its consideration around the world as a rare or mioritary disease, (low percentage of affected affected), therefore few grants are given for further exploration and research.

By putting together the subject of "Geriatric and infantile optometry", where we studied binocular dysfunctions and strabismus (either locomotor and accommodative), at the same time as the 5th International Congress of Glycogenosis, uniting forces, two students of the FOOT and a professor, Montserrat Augé i Serra, were at the Congress where we were lucky to be able to carry out a series of optometric examinations in patients affected with this disease thus having a large sample of patients with different subtypes of glycogen storage disease.

During 5-6-7th June 2014, in Bellaterra, the 5th International Congress of Glycogenosis took place, where a part from nutritionists and physicians attending, there were doctors specialized in liver and muscles. They were giving to each attendee a guide of conferences and two books about the effects on glycogen storage disease in Spain and new palliative therapies or others, discovered thanks to scientific research, apart from certain products as food for testing into the diets of the patients with Glycogenosis.

Vull donar les gràcies a totes les persones que m'han ajudat en el camí dels meus estudis, família sobretot, amics i parella, que amb les seves preguntes i ajuda, m'han portat a escriure aquest TFG amb moltes ganes i aspiracions futures per a estudiar més sobre un dels temes que tant m'apassiona, l'Optometria.

També voldria donar les gràcies al president de l'Associació Catalana d'afectats de Glucogenosis - Fransesc Cayuela - que ens ha permès fer aquest estudi en un congres tant important com aquest fent així possible un gran recull de dades per el nostre estudi.

Donar gràcies al president de l'AEEG (Asociación Española de Enfermos de Glucogenosis) - Alberto Molares-.

A l'hotel Campus UAB per cedir-nos una sala de dimensions suficients per a fer els exàmens.

La FOOT (Facultat d'Òptica i Optometria de Terrassa) per a cedir-nos els instruments.

Finalment donar les gràcies a la Professora Montserrat Augé i Serra per a deixar-nos col·laborar en un projecte tant ambiciós com aquest que mai s'havia dut a terme.

A tots vosaltres,
gràcies per fer-ho possible.

.

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1. Introduction

In the times and society in which we are currently living, life is elongated for more years than it used to before (Christensen K et al., 2010), but which is the quality of our living expectancy? Does more years equals to more quality of life? We do need support to find out better ways to life.

This is why science and new technologies are playing an important role, and that is why I wanted to do a study just for a minority disease, because main of the investigations are for big and epidemic diseases but minority illnesses as Glycogenosis are left behind with less economic support either for research and treatment, at least in a national reference.

Despite of the lower income of money for the global investigation of minority diseases, the patients suffering the illness are grouped together in associations were they can find psychological and medical advices in how to treat it. This is the case of the "*Asociación Española de Enfermos de Glucogenosis"* and where our project takes place in the "*V Congreso de Enfermos de Glucogenosis. 2014"* located in le UAB (*Universitat Autonoma de Barcelona*) where two students and the professor Montserrat Augé i Serra did some visual screening tests with the consent of the patients in order to find out if there was or was not any conclusive refractive or motility problems in the patients affected by Glycogenosis.

Taking into account that Glycogenosis is a muscular disease, the purpose of our study was to do a research in order to find out if the people affected with the Glycogenosis subtypes (Ia/Ib, II, IIIa and V), had any general visual problem, if any subtype of the malady had a distinctive or comparative trait or if the subtypes had any comparison in general, in a visual level whether it is orientation of visual axis, phorias or tropias, motility alterations, tracking movements and saccade eye movements.

2. Theoretical framework

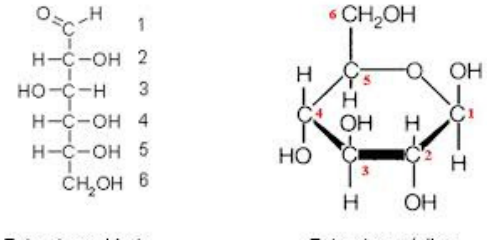
A) Glycogenosis

"Any of the glycogen deposition disease characterized by accumulation of glycogen of normal to abnormal chemical structure in tissue; there may be enlargement of the liver, heart, or striated muscle, including the tongue, with progressive muscular weakness. Seven types (Cori classification) are recognised, depending on the enzyme deficiency involved, all of autosomal recessive inheritance, but with a different gene for each enzyme deficiency."

(MIM designations: I,*232200, *232220, *232240; II, *232300; III, *232400; IV, *232500: V, *232600: VI, *232700: VII, *232800), (Mendelian Inheritance in Man))

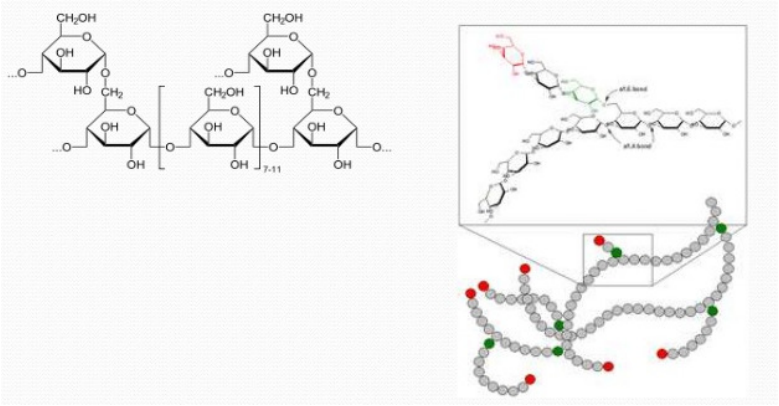
Glycogenosis is a group of rare diseases with a direct impact on Glycogen metabolism.

Glycogen is a molecule (polysaccharide) formed by subunits of Glucose (monosaccharides).



1. Glucose structure

Glucose is a kind of sugar (Carbohydrates) that our body absorbs from food like fruits or honey through our small gut and going to the bloodstream providing us energy. The organs in which Glucose is stored as Glycogen are the muscles and the liver.



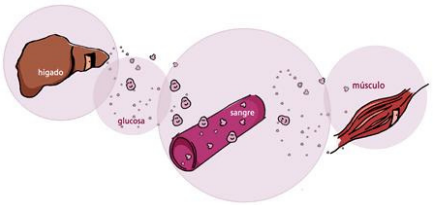
2. Glycogen chain.

Depending which is the Glycogen alteration we divide the disease in 3 types;

1. Muscular Glycogenosis.
2. Hepatic Glycogenosis.
3. General Glycogenosis (muscular, hapatic and cardio manifestations).

In each of the different types of Glycogenosis, we have some premises;

- Maintain blood Glycaemia levels around (80-130mg/ld. in children and 60-110mh/ld. in adults), having slow absorption HC meals every 2-4h.
- Avoid the increase of lactic acid and fatty acids in the bloodstream.



1. Glycose from liver to muscle.

Maintaining a strict diet, as;

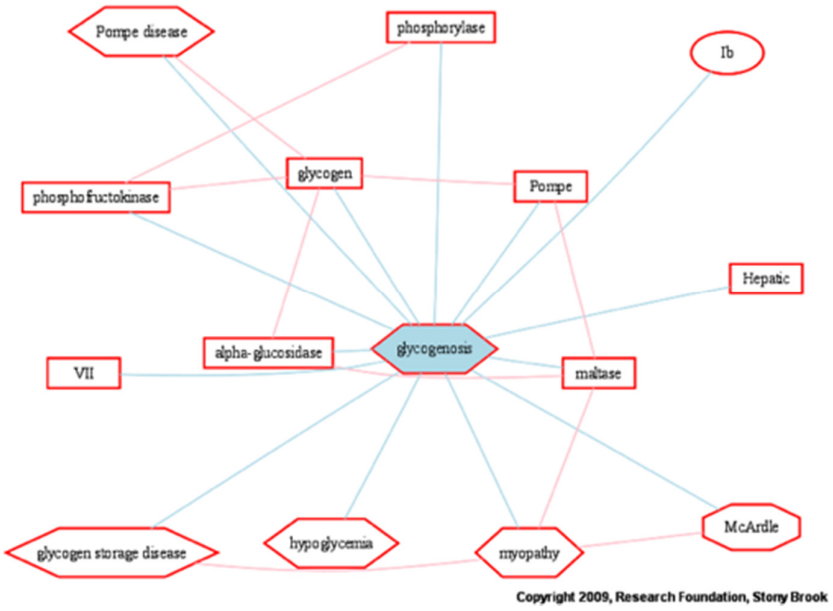
- Slow absorption Carbohydrates (60-70% daily energy).
- Fat (20-25% daily energy).
- Proteins (10-15% daily energy).
- Reduce lactose intake.
- Reduce the intake of fast absorption Carbohydrates.
- Reduce the intake of raw wheat.

4. Diet

A.1. Types

There is 8 different types of Glycogenosis;

1. Glycogenosis type I – Von Gierke disease
2. Glycogenosis type II – Pompe disease
3. Glycogenosis type III – Cori-Forbes disease
4. Glycogenosis type IV – Andersen disease
5. Glycogenosis type V – McArdle disease
6. Glycogenosis type VI – Hers disease
7. Glycogenosis type VII – Tarui disease
8. Glycogenosis type XI – Fanconi-Bickel



5. Different types of Glycogenosis and it's enzymes

In this project we are just going to mention 5 types of Glycogenosis, which are the ones we tested patients with.

There is an attached table in the annex part, explaining in a structured summary table all the features of each subtype of the disease.

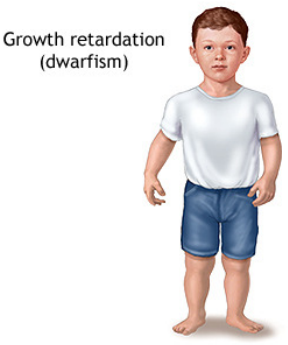
1) GLYCOGENOSIS TYPE Ia/Ib

(VON GIERKE ILLNESS) GSD1A /GSD1B

Ia)

It is known to be an hepatic disease, in which there is almost none or low functionality of the enzyme transforming Glucose-6-fosfate (G6P) to Glucose, or the enzyme in charge of transporting G6P into the endoplasmic reticulum, glucose-6-fosfate translocase (TG6P). It manifests during the first year of life with severe hypoglycaemia and hepatomegaly because of the Glycogen accumulation.

In both cases (Ia/Ib) Glucose synthesis and the obtaining of glucose by glycogen degradation is interrupted, so hepatic glucose is insufficient or there is none. Therefore the affected individuals exhibit delayed puberty, growth retardation, high levels of lactate (lactic academia) and fatty acids in blood (hyperlipidaemia), Glucose levels are diminished, and in adults a high incidence of hepatic adenomas. (Lei. K et al., 1993).



6. Growth retardation

Ib)

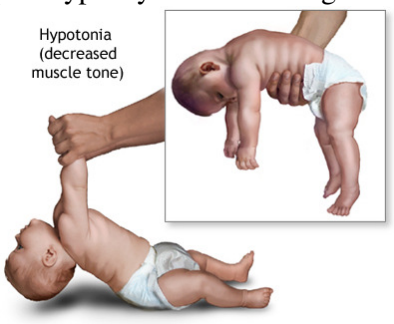
It is caused by a compound heterozygotes mutation or homozygotes in the G6PT1 gene, which encoding the translocase glucose-6-phosphate enzyme, in the 11q23 chromosome.

There has been a theory proposing the existence of a second type of Von Gierke made by (Senior et Loridan, 1968), saying that even the activity of G6PC (glucose-6-phosphatase) was present on in a vitro assay, glucose was not liberated from glucose-6-phosphate in vivo, which they had name it as – functional deficiency of G6P-. The affected individuals had shown the same symptoms as the type Ia Glycogenosis apart from having impaired enzyme function despite normal activity in homogenates.

2) GLYCOGENOSIS TYPE II

(POMPE DISEASE) GSD2

Known to be a general autosomal recessive disorder subtype of Glycogenosis, also known as prototypic lysosomal storage disease, is a subtype caused by an increasing accumulation of Glycogen inside the Lysosome. This is caused by a lack of the enzyme in charge of decomposing Glycogen to Glucose molecules (alfa-(1-4)-glycosidase aka acid maltase). This is interfering with the cellular function harming the cell, producing cardiomyopathy and muscular hypotonia in children, and in juvenile and adult forms there are the same symptoms plus involvement of skeletal muscles as the major sign. (Matsuishi et al., 1984).



7. Hypotonia

3) GLYCOGENOSIS TYPE IIIa

(CORI-FORBES DISEASE) GSD3

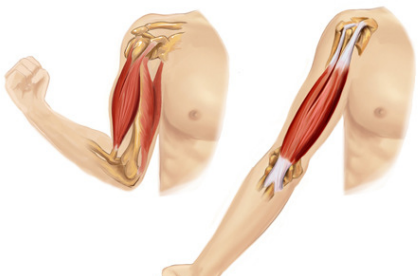
Glycogen storage disease III is a subtype of Glycogenosis (hepatic), due to a short chain accumulation of Glycogen inside the cells. It is an autosomal recessive metabolic disorder caused by a non or low activity of the debrancher enzyme A1,6G (amilo 1.6-glucosidase) which does not finish the process of Glucose degradation, and it is associated with an accumulation of abnormal glycogen with short chains.

The patients suffering from this subtype they often have the same symptoms as subtype I, but in a mild way. Most of them have the enzyme deficiency in the muscle and liver as subtype IIIa, and about the 15% are having deficiency just in the liver (subtype IIIb). (Shen et al., 1996). Especially in infancy or early childhood we can see signs as growth retardation, hepatomegaly and hypoglycaemia, muscle weakness can become severe in adults developing in rare cases cardiomyopathy. (Shen et al., 1996).

4) GLYCOGENOSIS TYPE V

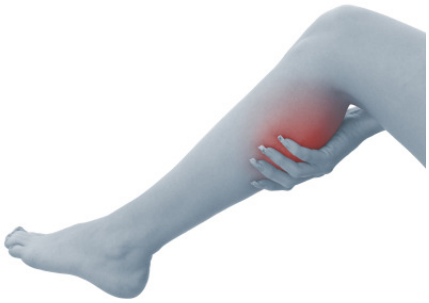
(McARDLE DISEASE) GSD5

McArdle disease is one of the muscular-autosomal-recessive-metabolic-disorder subtype of Glycogenosis, caused by an insufficient or non-enzyme activity (Phosphorylase). Because of the Glycogen decomposition in the liver, in order to provide Glucose (energy) to the rest of the organism, there's a low increase of Glycogen in the muscle which is after all used ask energy for contracting.



8. Muscular contraction

Due to the non-decomposing-Glycogen, the body reacts with weakness and skeletal muscular cramps during exercise which has intolerance on kids for being more physically active in childhood and adolescence, feeling muscle weakness, myalgia, and lack of endurance. Transient myoglobinuria may occur after exercise, due to rhabdomyolysis. Severe myoglobinuria may lead to acute renal failure.



9. Muscular cramps

McArdle disease is a benign disorder, except for the possibility of having a renal failure as a complication. (Chen, 2001).

3. Targets

Our main point of the study research was to evaluate and detect any visual dysfunction in the patients diagnosed with the glycogenosis subtypes mentioned above (Ia/Ib, II, IIIa,V).

Evaluating all the possible areas of the optometric exam, either from both points of view objectively and subjectively.

4. Methodology

A. Subjects

This examination has been done during de "V Congreso de Enfermos de Glucogenosis" a congress where all of patients attending were both, affected or familiars of the affected patients itself.
The subjects evaluated were first listed down, accepting afterwards the ethc consent of using the results of the optometric exam for a study.

In any case we rejected subjects, we tested everyone even if they were not having the disease, males and females, adults and kids, and this means that from all the subjects tested just 25 were useful for our research.

From 37 subjects evaluated, 26 were having Glycogenosis, 1 of the affected patients did not have his disease diagnosed in ay subtype, this is why we haven't add him in our analysis.

The Congress took place in the Autonomic University of Barcelona in Bellaterra, during four days 4-7th June 2014.

B. Process and optometric instruments

The procedure was the following, two volunteers, students of the Faculty of Optics and Optometry in Terrassa – FOOT, supervised by a teacher Montserrat Augé Serra, were invited to the congress in order to do visual exams to the attendants, in order to evaluate their visual system.

10. Volunteering and Catalan representative of AEEG

For each of the visual tests we have done, we have gathered the visual results from the patients in each of the five Glycogenosis subtypes.

Glycogenosis types

1. Glycogenosis type Ia/Ib – Von Gierke disease
2. Glycogenosis type II – Pompe disease
3. Glycogenosis type III – Cori-Forbes disease
4. Glycogenosis type V – McArdle disease

From all the tests done, we've picked up the ones showed below in a box because the other ones had no statistic difference to prove a real and general measure to believe in from the same subtype.

- History and Symptoms
- Visual acuity (Ph)
- Focimetry
- Autorefraction
- Cover Test (CT)
    - Near Vision
    - Distance vision
- Phoria measurement (Maddox rot)
- Motility
- Saccade movements
- Simultaneous Perception (SP)
- Suppression
- Fusion
- Stereopsis (ST)
- Near Convergence Point (NCP)
- Push up test (Accommodation)
- Accommodation flexibility
- Colour (Ishihara)
- Pupils

11. Near Convergence test

- Motor deviation
- Sensorial deviation
- NCP
- Motility
- Saccades
- Sensorial fusion
- ST
- Colour (Ishihara)
- Pupils

12. Cover test

C) Test distribution

From the visual tests shown in the previous page, I have decided to choose some, as the most important ones, which are the ones I have used for the upcoming analysis.

Station 1;

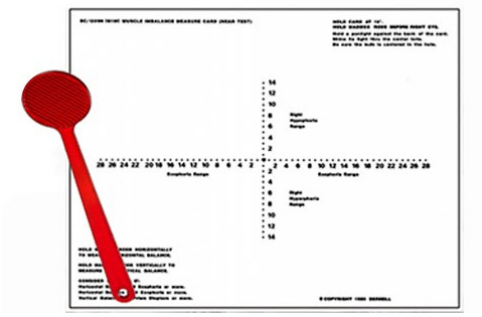
a) Motor deviation.

CT in a near position.
The subject is asked to look in a near target, where the specialist shows beforehand.
With the help of an occluder, the specialist observes if the eyes have any vertical or horizontal movement while covering and uncovering them in order to detect if there is a phoria, tropia or orthophoria.

Station 2;

b) Sensorial deviation

Phoria measured with Maddox root.
With the patient looking straight to a pen torch placed in the middle of the card (Figure 13), and holding a Maddox rot in front of his right eye, we ask whether the vertical/horizontal red line is located, and which the exact number is.



13. Phoria measurement card

c) Near Convergence Point (NCP)

14. RAF binocular gauge

The expert places the RAF binocular gauge in the patient's cheeks and it is asked to look at the accommodative target in front of his eyes, letters or numbers, as they should be seen clearly.
The specialist brings the target close to the patient's nose asking when the letters appear to be double. Once the target is double, the target is brought back in order to make the patient see it simple again. If the subject does not see double we write down GTN standing for Good To Nose or depending if there's a tropia and the patient was not able to fusionate.

d) Motility

The optometrist shows a target approximately at 40 cm asking the patient to look at the target following it with his eyes without moving his head, and saying if in any moment of the test there is pain, discomfort or double vision.
SPEC is written down if the eye movements are Soft Precise Extend and Complete.

e) Saccades

The subject has to keep his eyes in one of the two targets shown, alternating from target 1 to target 2 as the optometrists says so. The targets has a distance of 40 cm approx.
We write down if there is any fixation loss or regression.

f) Sensorial fusion

On a tablet and with the Application "Optonet", with a distance of 40 cm, Worth lights were shown in order to detect if the eyes were fusioning while the subject was wearing anaglyphic glasses with complimentary colours.

15. Sònia Travé doing a visual exam

Occluding one eye at a time, and the other and asking the patient how many lights could he see, where were them located, and which was the colour, and the same without occluding we can diagnose if the patient is having fusion or not, and see if he is supressing any of the eyes.

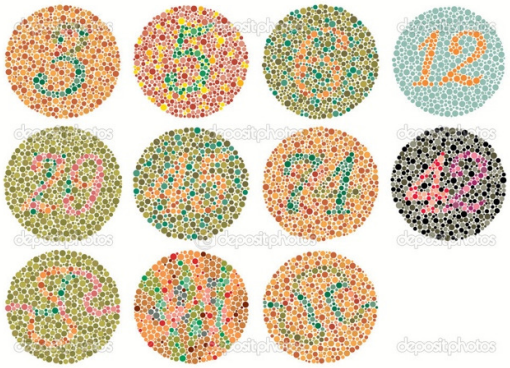
g) Stereopsis (ST)

With a TNO test held at 40 cm distance, plus anaglyphic glasses, the subject is asked to look at the stereoscopic plates.
The patient should see the different figures having a volume, getting closer or further away from the plate.

The plates with a value from 480 to 15 seconds of arc were presented until the subject was unable to identify three-dimensional shape correctly.

16. TNO test for stereoscopic vision

h) Colours (Ishihara)



17. Ishihara plates

Ishihara tests is a monocular test, so we have patched one eye of the patient, with the actual correction if they were wearing it, and proceed to ask for the numbers written in the different displayed plates.

In the plates there can be numbers or paths for the kids so it is easier for them to follow.

Any failing plates was written down as a mistake, and the failing colour.

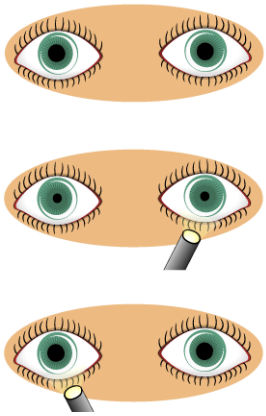
Colour Perception test was done using the Ishihara Isochromatic Cards, 24 Plates edition.

I) Pupils

We get the patient to look straight ahead, and we observe the pupil size and shape at rest, we also do observe the direct response (constriction of the illuminated pupil) once we flash a light with a pen torch, and then we observe the consensual response (constriction of the opposite pupil), repeating with the opposite eye.

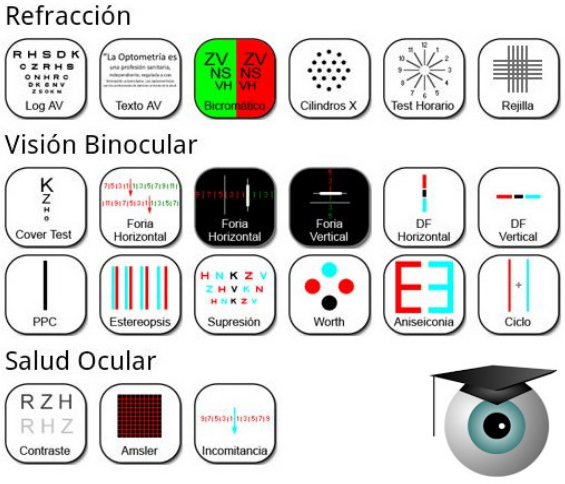
After that, we swing a light back and forth in front of the two pupils and compare the reaction to light stimuli.

Pupil reflexes tested were (direct, indirect and swing test).



18. Pupil swing test

For the near Application Optotypes for targets we have used the Bueno-Matilla (Optonet) as our visual tests



19. Bueno-Matilla tests

5. Statistical analysis

In the congress we had 37 subjects evaluated, from those, just 26 were having Glycogenosis, and 1 of the affected patients did not had a Glycogenosis type diagnosed, therefore in our study, we have picked up just the 25 affected subjects.

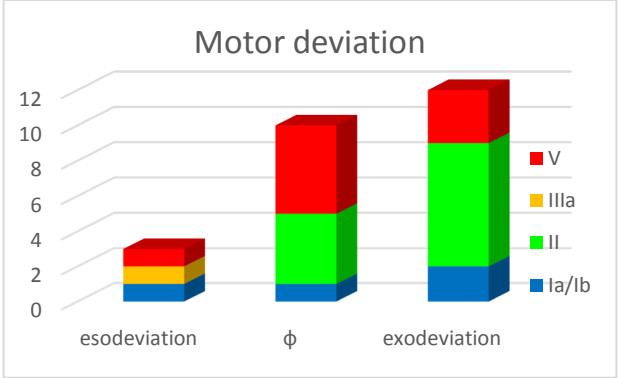
With subjects from 7 to 65 years old and having an average age of 35 years old, here we are showing some of the results.

Motor deviation for each subtype

+1 = Esodeviation (Either phoria or tropia).
0 = Orthodeviationa Φ
-1 = Exodeviation (Either phoria or tropia).

This measurement has been done doing CT in near, therefore, those are OBJECTIVE measures.

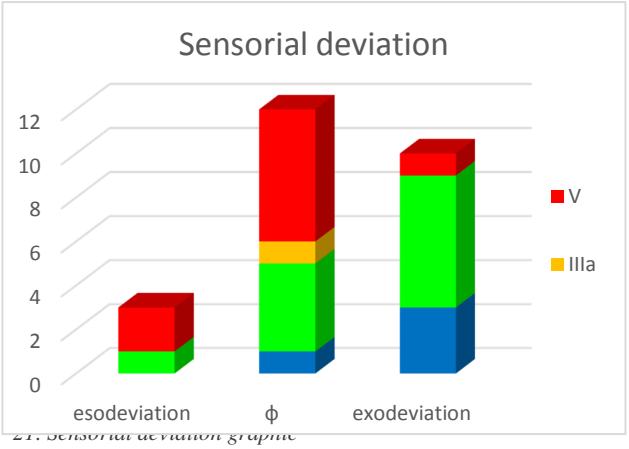
As we can see in the next graphic, 12 out of 25 patients tested are having an exodeviation, either by being a phoria or a tropia, which is almost a 50% of all the study, 48% to be precise.
Although the fact of having this divergent deviation there were some other cases minority 12% (3 cases), with Esodeviations, and the rest of the patients were having an Orthodeviation, 40% (10 cases).



20. Motor deviation graphic

Sensorial deviation

- = Exodeviation
+ = Esodeviation
0 = Orthodeviation

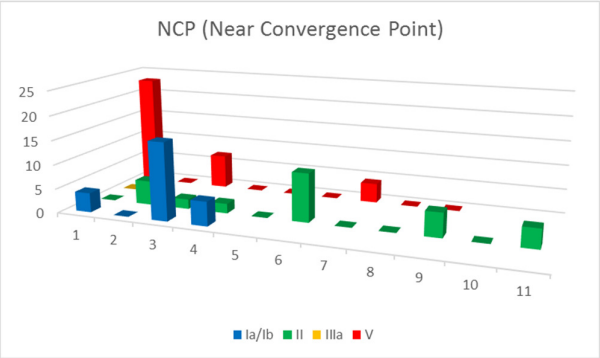


21. Sensorial deviation graphic

This graphic corroborates what we have seen in the previous one.
We are having more Exodeviations 40% (10 cases), in front or a 3 cases of esodeviation 12%, but with a increasing amount of Orthodeviations 48% (12 cases).
Even if there is less exodeviations and more orthodeviations than before, this is due to a the character of the test, one is a total Objective test, the practitioner decides whether or not there is a deviation and if it is affirmative, which kind it is, either convergent or divergent, and the sensorial deviation is totally Subjective, because it is the patient the one saying where he or she is seeing the line.

This measurement has been done with a Maddox rot and a Phoria measurement card, therefore those are SUBJECTIVE measures.

Near Convergence Point

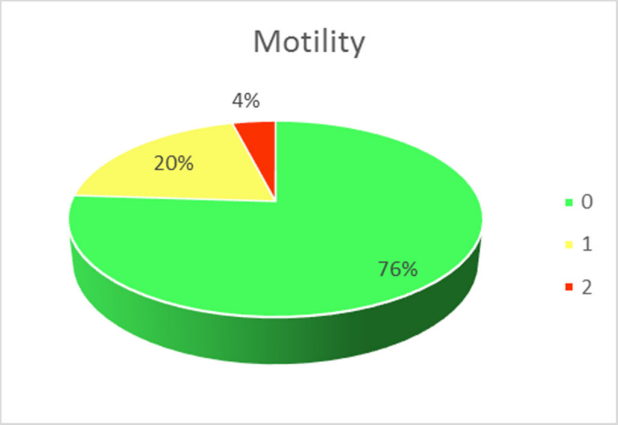


22. Near Convergence Point Graphic

Instead of showing the break-up and recover position, there is just the breaking-up distance measured in cm.
As shown in the graphic below, we can notice that up to a 52% (32 cases), can have thier eyes converging up to their noses GTN, in the second position we are having 10 cases (40%) that cannot converge up to their nose but they are within the normal values (5-10 cm break), and in the third position, we are having the 8% (2 cases), having theirs break up distance quite far away from the normal expectancy, 16 and 23 cm far.
There is no abnormal values apart from the tropias where it is quite ovious that NCP is not going to have a normal value, either because of the supression or because of the strenght of the muscle in order to converge.

Motility

0 = No affectation, everything is normal (S.P.E.C.).
1 = Affecatation in ¼ of the S.P.E.C. components, whosoever of them.
2 = Affecatation in 2/4 of the S.P.E.C. components, whosoever of them.
3 = Affecatation in 3/4 of the S.P.E.C. components, whosoever of them.
4 = Total affecatation, 4/4 of the S.P.E.C. components, whosoever of them.

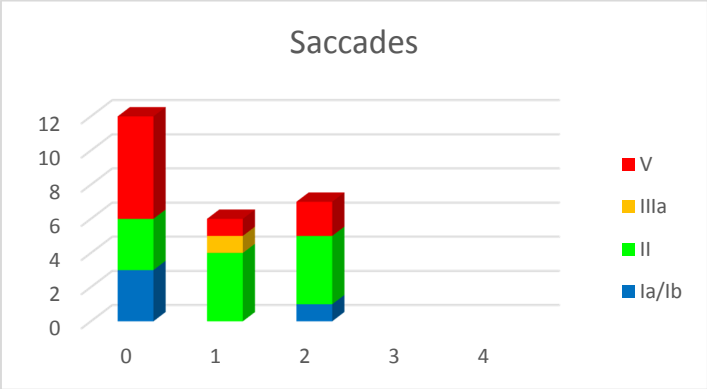


As we can see in the graphic in our left about a 76% of the subjects, 19 cases, are having no problem at all with any of the components of the motility test (Soft. Precise. Extend. Complete), but 5 cases are having trouble in any of the 4 compounds, and 1 case the subject is failing 2 out of four of the motility components, which is one of the subjects having a divergent tropia.

23. Motility graphics

Saccades

0 = No affectation, everything is normal (S.P.E.C.).
1 = Affecatation in ¼ of the S.P.E.C. components, whosoever of them.
2 = Affecatation in 2/4 of the S.P.E.C. components, whosoever of them.
3 = Affecatation in 3/4 of the S.P.E.C. components, whosoever of them.
4 = Total affecatation, 4/4 of the S.P.E.C. components, whosoever of them.

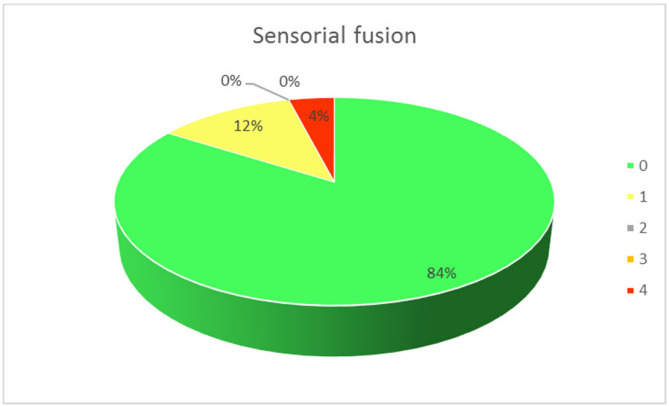


24. Saccades graphic

Introducing the graphics of saccade movements, and having the same reference a the graphic shown before, we can see that about 48% (12 cases) have no affectation, but about ¼ of the missing, struggle fixating in any of the coponents of S.P.E.C., and another 25% is having problems with at least 2 components at the time, this can be due to the muscle weakness inflicted by the pathology.

Sensorial fusion

0 = Stable fusion
1 = Intermittent fusion
2 = Intermittent suppression
3 = Constant suppression
4 = Diplopia



25. Sensorial fusion graphic

The provided information that the graphic is showing, is basically saying that most of the subject were having stable fusion 21 cases (84%) as that should be the normal visual status.

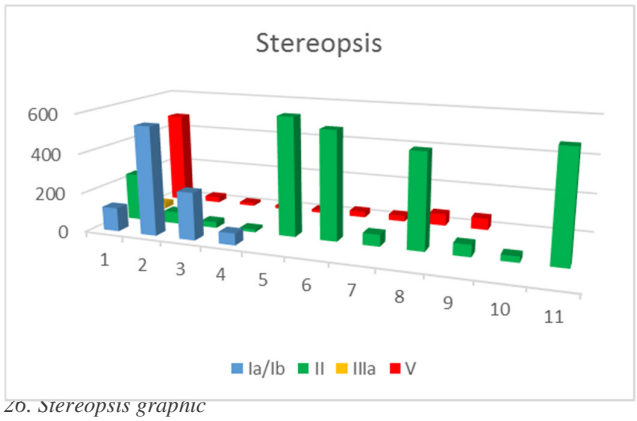
On the other side, we find out that there were 3 cases with intermittent fusion, this means that when we were dissociating the eyes there was a rupture of the visual axis causing a prompt intermittent diplopia.

There was just 1 case having Diplopia, in which all the other visual tests were altered.
Two out of the four subjects having problems fusioning were from the Glycogenosis type V.

Stereopsis (ST)

Last number of the vectograms seen in the TNO test.

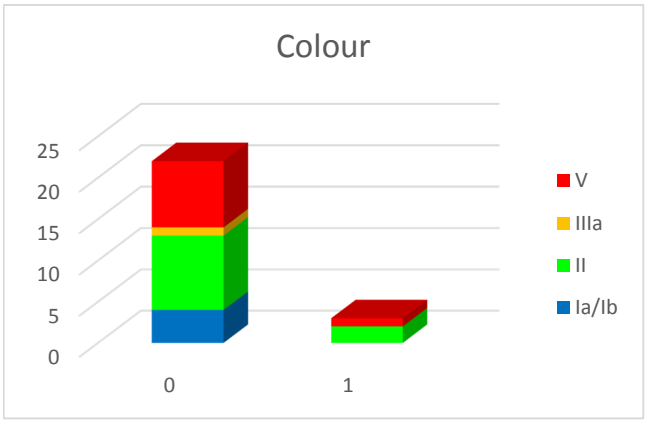
In this test we have had diversity in the answers, but up to 68% were having "normal" values (15"-120") for their respective ages, the remaining 32% had a lower level of stereocuty (>240"), which can be due to the decomensated phorias, uncorrected vision (just 1 case) or even tropias.



26. Stereopsis graphic

Colour

0 = No affectation
1 = 1 or more mistakes



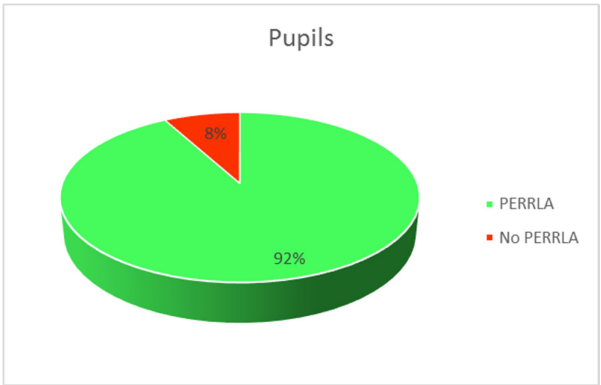
27. Colour graphic

In this test, we have just found 3 out of 25 subjects having a colour anomaly.
2 of them were from the Pompe subtype, and the other one was from the McArdle.
No conclusive results have been found in this test, as colour blindness is inherited, and the affected subjects were males, which have more predisposition to be colorblind. "8% of males and 0.5% of females are colorblind" (Deeb, 2005).

Pupils

0 = Pupils not affected (PERRLA)
1 = Any of the components of PERRLA affected.

As the graphic is showing in our right, Glycogenosis has nothing to see with the pupils and it's affereance/effeareance.
We just find out that 2 of the patient's pupils were not round because of ocular surgeries.



28. Pupils graphic

6. Conclusions

Taking into account all of the mentioned above, we can summarise all saying that we find out that most of the tested subjects affected with Glycogenosis are having exodeviations, both, as motor and sensorial, and thus, being able to affect NCP and, Motility.
This does not jeopardize the visual system, even if in the Saccades, about half of the tested subjects have had a fixation problem, (can be due to divergent deviations).

Talking about the Stereopsis results we can say that they have been pretty standards for the respective patient's ages that can be because most of the deviations were phorias and not tropias, except for two of them, were they had the lowest stereoacuty of the TNO test. This was due to the suppression or diplopia in the different mentioned cases.

Apart from that, Colours and pupils results were not showing abnormal results, most of the patients were not having any problem with the tests.

All the ocular motility that implies moving the eyes as one, from one point to another, or following, was expected to have abnormal values, because Glycogenosis implies having muscle weakness all over the body inflicted by the pathology, which affects also the eyes movements, and as we have seen more in the type II, followed by type V.

(The conclusive values are obtained through the optometric tests, explained on this project, and the conclusions of each graphic were extracted based on the normal values for each person's age).

7. Limitations & future prospects

We had a sample of 25 subjects, but a future study with higher amount of subjects would make a study more complete.
For next optometrist researching in this fields I would strongly advise testing people with the disease but at the same time having a control group so they would know the current differences, being able to have a more accurate study with real findings.

For the nice patients we had there is a bright future upon them, because new technologies as enzyme replacement are coming "Enzyme replacement therapy (ERT) with human recombinant alpha-glucosidase (rhGAA) has been demonstrated to be effective in the treatment of infantile forms", more diets are being developed and more high tech research is being done in this field. (Bembi B., 2010) & (Léon P. F., 2003).

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- (MIM designations: I,*232200, *232220, *232240; II, *232300; III, *232400; IV, *232500: V, *232600: VI, *232700: VII, *232800), (Mendelian Inheritance in Man)

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ANNEX

1) Source of the images

- Figure 1. Glucose structure: https://lookfordiagnosis.com/mesh_info.php?term=glucose-6-phosphate&lang=1
- Figure 2. Glycogen chain: https://en.wikibooks.org/wiki/Principles_of_Biochemistry/Gluconeogenesis_and_Glycogenesis
- Figure 3. Glycose from liver to muscle: http://healthy-ojas.com/diabetes/diabetes-organs.html
- Figure 4. Diet: http://balanceddietelisa.blogspot.com.es/2015/09/nutrition-and-balanced-diet.html
- Figure 5. Different types of Glycogenosis and it's enzymes: http://www.textmed.com/disease/glycogenosis.htm
- Figure 6. Growth retardation: http://www.pennmedicine.org/encyclopedia/em_PrintPresentation.aspx?gcid=100146&ptid=3
- Figure 7. Hypotonia: https://www.nlm.nih.gov/medlineplus/ency/imagepages/17229.htm
- Figure 8. Muscular contraction: http://jonbarron.org/article/physiology-muscles
- Figure 9. Muscular cramps: https://www.antoniopacelli.com/community/article/leg-cramps-and-stiches
- Figure 10. Volunteerings and Catalan representative of AEEG: http://www.coooc.cat/noticia.asp?id_noticia=515
- Figure 11. Near convergence point: http://www.bernell.com/product/3146/1250
- Figure 12. Cover test: http://www.convergenceinsufficiency.net/detail.asp?id=18&pid=13
- Figure 13. Phoria measurement card: https://i.ytimg.com/vi/2RYJmLQGrZ0/hqdefault.jpg
- Figure 14. RAF binocular gauge: http://www.bocinstruments.com.au/shop/item/clement-clarke-raf-binocular-gauge/clement-clarke

- Figure 15. Sònia Travé doing a visual exam.
  http://www.coooc.cat/noticia.asp?id_noticia=515

- Figure 16. TNO test for stereoscopic vision:
  http://www.graftonoptical.com/products/775-tno-stereo-test.html

- Figure 17. Ishihara plates:
  http://sp.depositphotos.com/6776121/stock-illustration-complete-ishihara-color-test-plates.html

- Figure 18. Pupil swing test:
  http://stanfordmedicine25.stanford.edu/the25/pupillary.html

- Figure 19. Bueno-Matilla test:
  http://www.1mobile.es/optonet-remote-1-0-196379.html

- Figure 20. Motor deviation graphic

- Figure 21. Sensorial deviation graphic.

- Figure 22. Near Convergence test graphic

- Figure 23. Motility graphic

- Figure 24. Saccades graphic

- Figure 25. Sensorial fusion graphic

- Figure 26. Stereopsis graphic

- Figure 27. Colour graphic

- Figure 28. Pupils graphic

2) Consent letter

Benvolgudes famílies,

L'informem del desenvolupament d'un estudi que estem duen a terme per a la detecció de l'afectació de Glucogenosis a nivell visual.

L'objectiu és valorar mitjançant tests de refracció, binocularitat i acomodació la normalitat dels sistemes visuals en pacients afectats amb aquesta malaltia.

Els controls visuals que es realitzaran en les instal·lacions Hotel Campus de Bellaterra en els 5,6,7 de Juny de 2014 i hores donades segons inscripció en llista, són totalment gratuïts i aniran acompanyades d'un informe complert per les famílies.

Esperant que en traieu profit, us donem les gràcies per la vostra col·laboració.

Jo, ... com a pare/mare o tutor de ....., amb DNI ....., dono el meu consentiment a que es faci un control visual al meu fill/filla .....

Aquest controls visuals en aquest congres forma part d'un projecte de final de grau en òptica i optometria que té com ha objectiu valorar l'afectació visual en pacients afectats amb glucogenosis.

Segons el que estableix la Llei Orgànica de Protecció de Dades de Caràcter Personal, l'informem que el tractament de dades personals dels seu fill/filla serà específicament per al projecte final de Grau amb finalitat sanitària i docent.

Per tant, dono el meu consentiment voluntari per a realitzar les proves i preguntes necessàries per realitzar l'estudi.

Atentament, Firma de consentiment

Sònia Travé Huarte

UNIVERSITAT POLITÈCNICA DE CATALUNYA
BARCELONATECH
Facultat d'Òptica i Optometria de Terrassa

3) Optometric test file

NOM I COGNOMS:	
DATA DE NAIXEMENT:	EDAT:
ESCOLA:	CURS:

ESTAT REFRACTIU Rx: OD: AV:

Usuari d'ulleres □NO □SI OI: AV:

AVsc	OD:	pH:	OI:	pH:	AO:
RETINOSCOPÍA	OD:				
	OI:				
Sx	OD:		AVcc:		
	OI:		AVcc:		

VISIÓ BINOCULAR I ACOMODACIÓ **Totes les proves amb la correcció habitual**

CT VL: □Fòria □Tròpia □Φ □X □E □OD □OI □ Intermitent □Alternant

CT VP: □Fòria □Tròpia □Φ □X □E □OD □OI □ Intermitent □Alternant SEGUIMENTS: S P E C SACÀDICS: S P E C

PERCEPCIÓ SIMULTÀNIA: □SI □NO

SUPRESSIÓ: □NO □ SI: □TOTAL □ALTERNANT □INTERMITENT □OD □OI

FUSIÓ: □ SI □NO □ESTABLE □INESTABLE

ESTEREÒPSIA: PPC (R/r):

DOMINÀNCIA OCULAR: MOTORA □OD □OI SENSORIAL □OD □OI

FORIA VP: RESERVES: BN: BT:

FV VP:BN: BT:

PPA: AAOD: AAOI:

4) Symptom questionnaire

QUESTIONARI DE SIMPTOMES

Nom i cognoms.....

SIMPTOMES (Marcar amb una X el requadre corresponent)	SI	A vegades	NO
1. Em canso quan porto una estona mirant de prop			
2. Em fa mal el cap quan porto una estona llegint			
3. Veig borrós quan intento llegir			
4. Quan llegeixo, veig doble			
5. Quan llegeixo, em ploren els ulls			
6. Quan llegeixo em costa concentrar-me			
7. Quan llegeixo, noto que es mouen les lletres, les paraules o les línees			
8. Quan llegeixo, m'agafa son			
9. Quan porto una estona llegint, em costa més entendre el que llegeixo			
10. Llegeixo massa lentament			
11. Crec que giro un ull al llegir			
12. Tanco un ull per veure millor			
13. Sento tensió als ulls quan estic mirant alguna cosa una estona			
14. Quan llegeixo una estona, em distrec amb facilitat			
15. M´acosto o allunyo molt per llegir			
16. Tinc de moure el cap per poder llegir			
17. Em perdo quan estic llegint			
18. Quan llegeixo, em salto algunes paraules o línees			
19. Em resulta difícil copiar de la pissarra			
20. Freqüentment em fa mal el cap			
21. Tinc dificultat per mirar de la pissarra a la llibreta i al reves			
22. Em molesta molt la llum			
23. Sento que em cremen els ulls al llegir			

5) Symptom table

How the table Works:

A)

- I get tired while looking near.
- When I am reading my eyes tear.
- It is hard for me to focus while reading.
- I feel a burning sensation while reading
- I have eye strain while reading.
- After reading for a while I get distracted.

B)

- When I am trying to read everything is blurry.
- Distance vision is blurry.

C)

- I see double while reading.

D)

- I am having headaches frequently.

E)

- I am very light sensitive.

F)

- I am closing one eye to see better.

G)

- I get either really close or really distant to read.

	Symptoms	A	B	C	D	F	G
Ia	1	0	0	0	0,5	0,5	0
	2			0	0	0	1
	3	0	0	0	0	0	0
Ib	1	0	0	0	0	0	0
II	1						
	2	0	0	0	0	0	0
	3		0	0	0	0	0
	4				0,5	0,5	0
	5		0	0	0,5	0	0
	6		0,5	0	0	0	0
	7				0	0	0,5
	8	0	0	0	0	0	0
	9	0		0	0	0	0
	10	0		0	0,5	0	0
	11					0	0
IIIa	1			0	0	0	1
V	1		0	0	1	0	0
	2	0	0	0	0,5	0	0
	3	0	0	0	0	0	0
	4	0	0	0	0	0	0
	5		0	0		0	0
	6		0	0	0	0	0,5
	7	0		0	0	0	0
	8	0	0	0	0,5	0	0
	9	0	0	0,5	0	0	1

0 Meaning NO/NEVER

0.5 Meaning Sometimes

1 Meaning YES/ALWAYS

6) Patient feedback

	Visió Binocular								Acomodació						
	CT		Seguiments	Sacàdics	PS	Supressió	Fusió	Estereòpsia	PPA		FA bino VP	FA		Color (Ishihara)	Pupil·les
	VL	VP							AA_{UD}	AA_{UE}		VP_{UD}	VP_{UE}		
Tipus															
Ia															
Ib															
II															
III															
V															

Fisiopatología hepàtica hipoglucèmica

Fisiopatología muscular

Fisiopatología peculiar

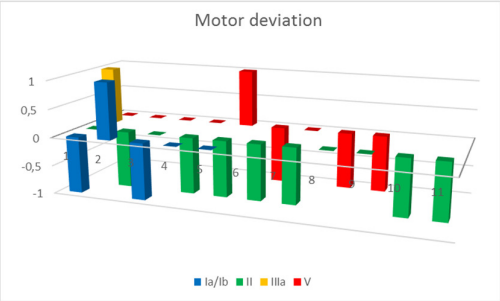
7) Patient results

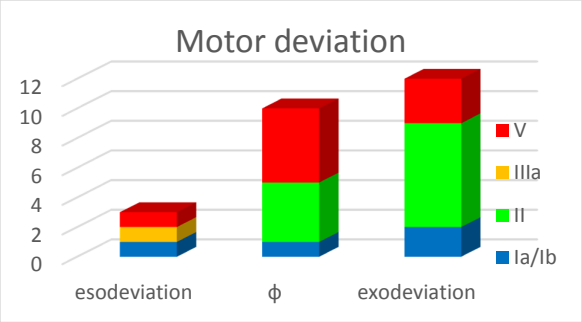
Type		Gender	Age	Motor deviation	Sensorial deviation	NPC	Motility	Sacades	Sensorial fusion	ST	Colour (Ishihara)	Pupils
Ia	1	1	36	-1	-10	4	0	0	1	120"	0	0
	2	1	7	1	-1	0	2	2	0	550"	0	0
	3	1	22	-1	-27	16	0	0	0	240"	0	0
Ib	1	1	41	0	0	5	0	0	0	60"	0	0
II	1	1	65	0	1	0	0	0	0	240"	0	0
	2	1	52	-1	-2	5	0	1	0	60"	1	0
	3	1	43	0	0	2	1	1	0	30"	1	0
	4	1	13	-1	-1	2	1	1	1	15"	0	0
	5	2	51	-1	0	0	0	2	0	600	0	0
	6	2	63	-1	-10	10	0	2	0	550"	0	0
	7	2	58	-1	-6	0	1	1	0	60"	0	0
	8	2	52	0	-6	0	0	2	0	480"	0	0
	9	2	60	0	0	5	1		0	60"	0	1
	10	2	56	-1	-4	0	0	0	0	30"	0	0
	11	1	6	-1	0	4	0	2	0	550"	0	0
IIIa	1	2	10	1	0	0	1	1	0	30"	0	0
V	1	2	32	0	-10	23		2	4	480"	0	1
	2	1	13	0	2	0	0	1	0	30"	0	0
	3	2	13	0	0	7	0	0	1	15"	0	0
	4	2	60	0	0	0	0	0	0	15"	0	0
	5	1	20	1	5	0	0	0	0	15""	0	0
	6	1	16	-1	0	0	0	0	0	30"	0	0
	7	1	14	0	0	4	0	0	0	30"	0	0
	8	1	62	-1	0	0	0	2	0	60"	1	0
	9	1	11	-1	0	0	0	0	0	60"	0	0

In deviations -1 = exodeviation 0 = orthodeviation +1 = esodeviation	In Motility & Saccades: 0 = No affectation S.P.E.C. 1 = Affectation in 1 component of S.P.E.C. 2 = Affectation in 2 components of S.P.E.C. 3 = Affectation in 3 components of S.P.E.C. 4 = Total affectation of S.P.E.C.
In Colour 0 = No affectation 1 = 1 or more mistakes	In Pupils 0 = Pupils not affected (PERRLA) 1 = Any of the components of PERRLA affected.

Motor deviation

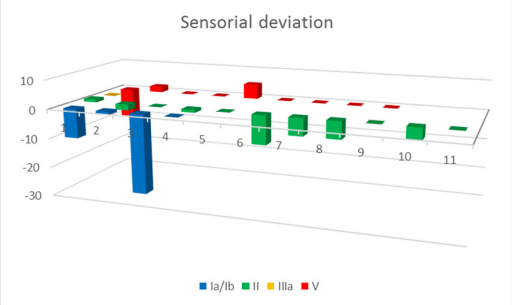
Motor dev		Ia/Ib	II	IIIa	V
	esodeviation	1		1	1
	ϕ	1	4		5
	exodeviation	2	7		3

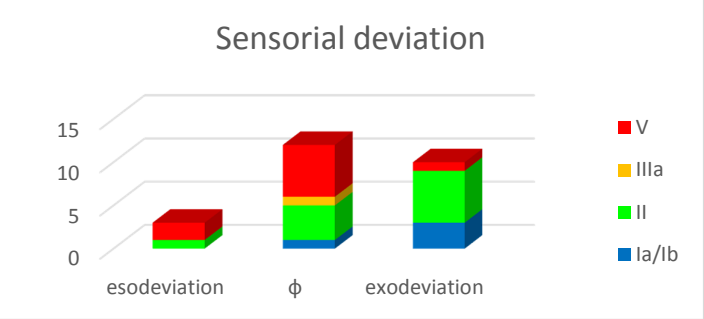




Sensorial deviation

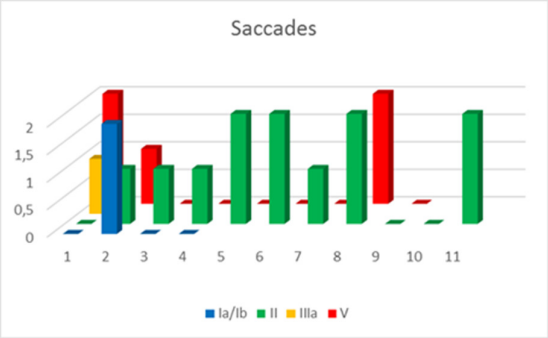
Sens dev		Ia/Ib	II	IIIa	V
	esodeviation		1		2
	ϕ	1	4	1	6
	exodeviation	3	6		1

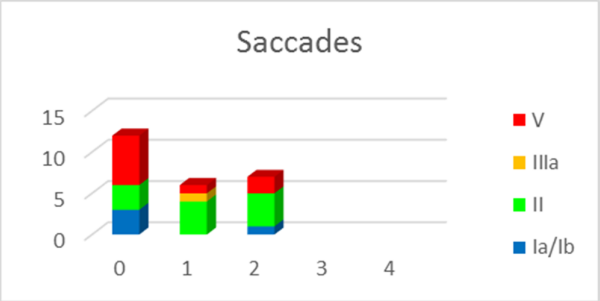




Saccades

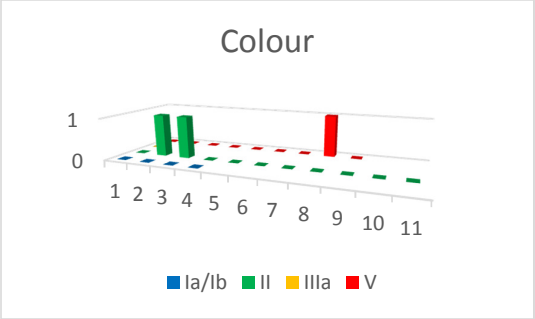
Saccades		Ia/Ib	II	IIIa	V
	0	3	3		6
	1		4	1	1
	2	1	4		2
	3				
	4				

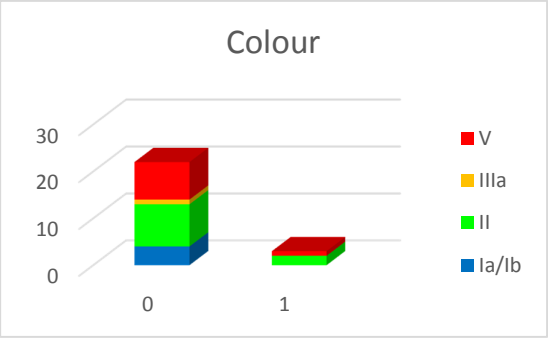




Colour

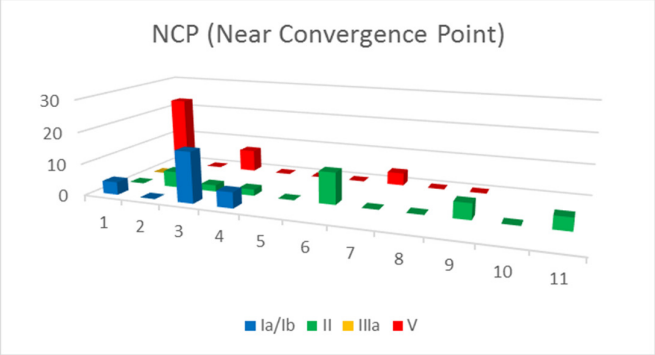
Colour		Ia/Ib	II	IIIa	V
	0	4	9	1	8
	1		2		1





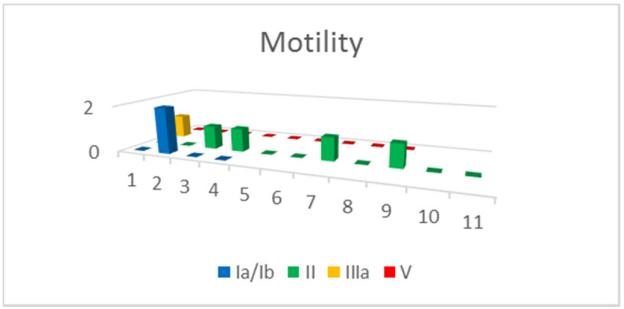
NCP

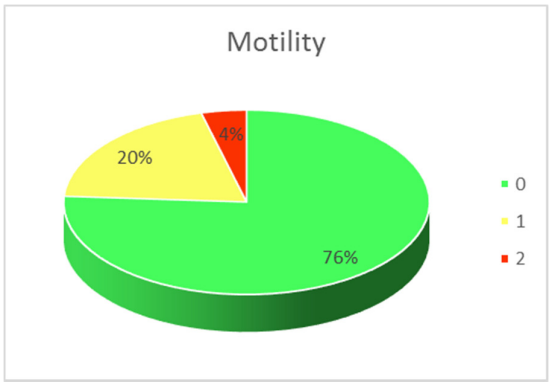
NCP	Ia/Ib	II	IIIa	V
	4	0	0	23
	0	5		0
	16	2		7
	5	2		0
		0		0
		10		0
		0		4
		0		0
		5		0
		0		
		4		



Motility

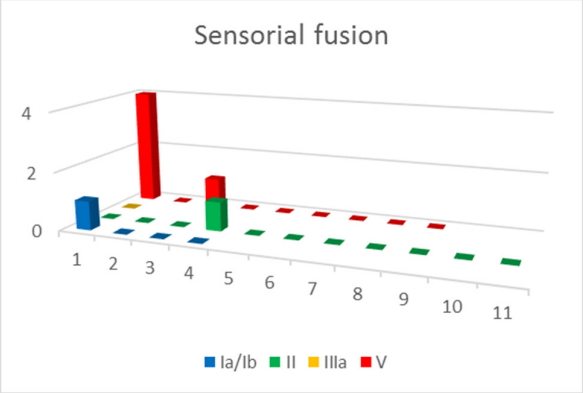
motility	Ia/Ib	II	IIIa	V
	0	0	1	0
	2	0		0
	0	1		0
	0	1		0
		0		0
		0		0
		1		0
		0		0
		1		0
		0		
		0		

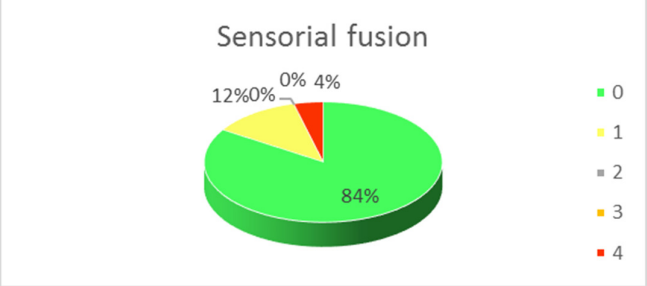




Sensorial fusion

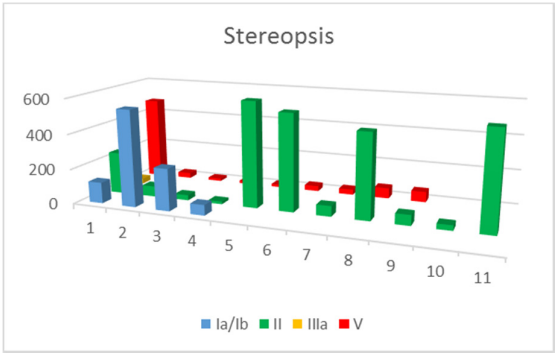
Sens fusion	Ia/Ib	II	IIIa	V
	1	0	0	4
	0	0		0
	0	0		1
	0	1		0
		0		0
		0		0
		0		0
		0		0
		0		0
		0		
		0		





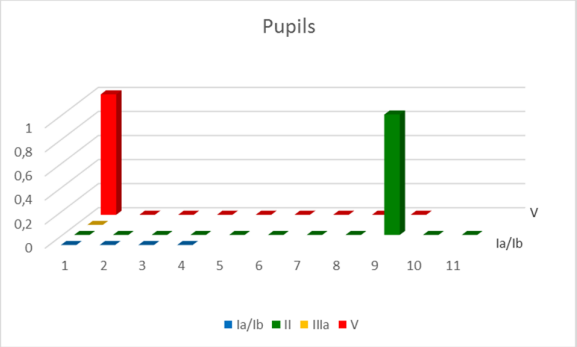
Stereopsis

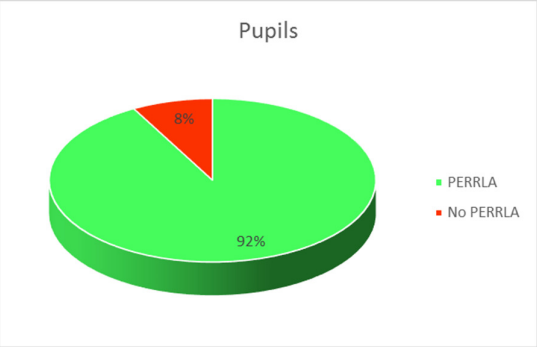
Stereopsis	Ia/Ib	II	IIIa	V
	120	240	30	480
	550	60		30
	240	30		15
	60	15		15
		600		15
		550		30
		60		30
		480		60
		60		60
		30		
		550		



Pupils

Pupils	Ia/Ib	II	IIIa	V
	0	0	0	1
	0	0		0
	0	0		0
	0	0		0
		0		0
		0		0
		0		0
		0		0
		1		0
		0		
		0		





GLYCOGEN STORAGE DISEASE Ia; GSD1A, (VON GIERKE ILLNESS)

CATEGORY	Subcategory	FEATURES	
Inheritance	-	Autosomal recessive	
Growth	Height	Short stature	
	Other	Delayed puberty	
Head and Neck	Face	'Doll-like' facies	
	Eyes	Lipemia retinalis	
Cardiovascular	Vascular	Hypertension	
Abdomen	External Features	Protuberant abdomen	
	Liver	Liver adenomas Hepatocellular carcinoma Hepatomegaly	
	Pancreas	Pancreatitis	
	Gastro intestinal	Intermittent diarrhea	
Genitourinary	Kidneys	Reduced creatinine clearance	Renal stones Renal enlargement
Skeletal	-	Osteoporosis Gouty arthritis	
Skin, Nails, Hair	Skin	Xanthoma	
Muscle, Soft Tissue	-	Decreased muscle mass	
Metabolic Features	-	Hypoglycemia	
Hematology	-	Bleeding diathesis	
Laboratory Abnormalities	-	Glucose-6-phosphate deficiency Hyperlipidemia Hyperuricemia Lactic acidosis	Hypoglycemia Proteinuria Liver transaminases normal to slightly increased
Miscellaneous	-	Often diagnosed between ages 3-4 months Early diagnosis and treatment prevent many complications	
Molecular Basis	-	Caused by mutation in the glucose-6-phosphatase, catalytic gene (G6PC)	

GLYCOGEN STORAGE DISEASE Ib; GSD1B (VON GIERKE ILLNESS)

CATEGORY	Subcategory	FEATURES
Inheritance	-	Autosomal recessive
Growth	Height	Short stature
	Other	Delayed puberty
Head and Neck	Face	'Doll-like' facies
	Eyes	Lipemia retinalis
	Mouth	Oral ulcers
Cardiovascular	Vascular	Hypertension
Abdomen	External features	Protuberant abdomen
	Liver	Hepatomegaly Liveradenomas Hepatocellular carcinoma
	Pancreas	Pancreatitis
	Gastro-intestinal	Chronic inflammatory bowel disease (IBD) Intestinal mucosal ulceration
Genitourinary	Kidneys	Reduced creatinine clearance Focal segmental glomerulosclerosis Renal stones Renal enlargement
Skeletal	-	Osteoporosis Gouty arthritis
Skin, Nails, Hair	Skin	Xanthoma
Hematology	-	Neutropenia Abnormal leukocyte function
Laboratory Abnormalities	-	T1 transport protein (Glucose-6-phosphate translocase) defect Hyperlipidemia Hyperuricemia Lactic acidosis Hypoglycemia Proteinuria Liver transaminases normal to slightly increased
Miscellaneous	-	Recurrent bacterial infections
Molecular Basis	-	Caused by mutation in the glucose-6-phosphate transporter 1 gene (G6PT1)

GLYCOGEN STORAGE DISEASE II; GSD2, (POMPE DISEASE)

CATEGORY	Subcategory	FEATURES	
Inheritance	-	Autosomal recessive	
Head and Neck	Ears	Hearing loss	
	Mouth	Macroglossia	
Cardiovascular	Heart	Cardiomegaly Shortened P-R interval on EKG	Huge QRS complexe Wolf-Parkinson-Whi syndrome
	Vascular	Cerebral artery aneurysm	
Respiratory	-	Respiratory failure due to muscle weakness Dyspnea Respiratory infections	
Chest	Ribs,Sternum, Clavicles and Scapulae	Diaphragmatic paralysis	
Abdomen	Liver	Hepatomegaly	
	Spleen	Splenomegaly	
Muscle, Soft Tissue	-	Weakness Proximal muscle weakness	Myopathic pattern on EMG Firm muscles
Neurologic	CNS	Hypotonia	Abnormal brain myelination
	PNS	Absent deep tendon reflexes	
Metabolic Features	-	Fever of central origin	
Laboratory Abnormalities	-	Elevated serum creatine kinase Elevated AST and LDH, especially infantile-onset	Presence of vacuoles on muscle biopsy Deficiency of alpha-1,4-glucosidase (acid maltase)
Miscellaneous	-	Two presentations - rapid, fatal disorder of infancy and slowly progressive muscular disorder of childhood Patients with later onset have better prognosis Incidence of 1 in 40,000 infants worldwide	
Molecular Basis	-	Caused by mutation in the alpha-1,4-glucosidase gene (GAA)	

GLYCOGEN STORAGE DISEASE III; GSD3, (CORI-FORBES DISEASE)

CATEGORY	SUBCATEGORY	FEATURES	
Inheritance	-	Autosomal recessive	
Growth	Height	Short stature	
	Other	Growth retardation	
Head and Neck	Face	Midface hypoplasia	
	Eyes	Deep-set eyes	
	Nose	Depressed nasal bridge Broad upturned nasal tip	
	Mouth	Bow-shaped lips Thin vermilion border	
Cardiovascular	Heart	Cardiomyopathy Ventricular hypertrophy on ECG	
Abdomen	Liver	Hepatomegaly Hepatic fibrosis	
Muscle, Soft Tissue	-	Muscle weakness (increases with age) Distal muscle wasting Myopathy Muscle biopsy shows vacuoles containing PAS-positive glycogen	
Metabolic Features	-	Hypoglycemia	
Laboratory Abnormalities	-	Amylo-1,6-glucosidase deficiency Hypoglycemia Hyperlipidemia Normal blood lactate	Normal uric acid Elevated transaminases Increased serum creatine kinase
Miscellaneous	-	Type IIIa has both liver and muscle involvement Type IIIb liver involvement only (15% of all cases)	Liver symptoms improve with age and disappear after puberty Muscle weakness increases with age
Molecular Basis	-	Caused by mutation in the amylo-1,6-glucosidase, 4-alpha-glucanotransferase gene (AGL)	

GLYCOGEN STORAGE DISEASE V; GSD5, (Mc ARDLE DISEASE)

CATEGORY	Subcategory	FEATURES
Inheritance	-	Autosomal recessive
Genitourinary	Kidneys	Myoglobinuria Dark urine following exercise
Muscle, Soft Tissue	-	Skeletal muscle weakness Decreased exercise capacity Muscle pain and cramps following exercise Rhabdomyolysis
Laboratory Abnormalities	-	Muscle glycogen phosphorylase deficiency Increased creatine kinase Increased ammonia with exercise Increased uric acid with exercise
Miscellaneous	-	Symptoms usually appear in adulthood 'Second wind' phenomenon Painful cramping following ischemic exercise test
Molecular Basis	-	Caused by mutation in the muscle glycogen phosphorylase gene (PYGM)

8) Optometric results

Podría tener un problema visual que interfiere en su rendimiento. Se recomienda un control visual en un gabinete optométrico.

En la revisión visual que hemos realizado en el Congreso de Glucogenosis de Junio 2014 hemos obtenido los siguientes valores:

Paciente	**Ojo derecho**	**Ojo izquierdo**
Agudeza visual de lejos		
Refracción ocular		
Motilidad ocular		
Binocularidad		
Visión del color		
Salud ocular		

Se recomienda que el optometrista revise las siguientes habilidades visuales:

	si	**no**
Agudeza visual de lejos		
Refracción ocular		
Motilidad ocular		
Acomodación		
Binocularidad		
Visión del color		
Salud ocular		

9) Programm for the 2014 Glycogenosis convention

Congreso año 2014 V CONGRESO INTERNACIONAL DE GLUCOGENOSIS (http://www.glucogenosis.org/v-congreso-internacional-de-glucogenosis/)

	Miércoles 4/06		
09:30 a 13:30 15:00 a 19:00	Evaluación Médica Glucogenosis Tipo II - Pompe		
	Jueves 5/06		
09:30 a 13:30 15:00 a 19:00	Evaluación Médica Glucogenosis Tipo II - Pompe		
09:30 a 13:30 18:00 a 20:00	Registro Nacional de Glucogenosis		
09:30 a 13:30 15:00 a 19:00	Revisión visión: Examen Optométrico (abierto para todos los tipos de Glucogenosis)		
16:00 a 18:00	Taller de Cocina con Manuel Román del Hospital Xeral de Vigo		
16:00 a 18:00	Visita Sincrotró Alba	**Salida desde Hotel Campus en autocar a las 15:30h*	
18:00 a 21:00	McGym (Gimnasia Glucogenosis Tipo V - McArdle)	**Lugar SAF (Servicio de Actividad Física de la UAB)*	
	Viernes 6/06		
09:30 a 13:30 15:00 a 19:00	Evaluación Médica Glucogenosis Tipo II - Pompe		
09:30	Recepción | Entrega de documentación		
10:00	Bienvenida V Congreso | Entrega reconocimiento		
10:30	Ponencia Jordi Duran		
11:00	Ponencia Federico Mingozzi y Giuseppe Ronzitti		
11:30	Pausa Café		
12:00	Ponencia Mercedes Serrano		
12:30	Mesa redonda: AEEG - Instituto de Salud Carlos III - Hospital Vall d'Hebron		
13:00	Visita Dep. Ingeniería Química UAB - Proyecto MELiSSA	**Hora prevista de llegada 15:30 - Salida desde Hotel Campus*	
13:00	Visita GTD Barcelona - Ingeniería Aeronáutica	**Hora prevista de llegada 15:30 - Salida desde Hotel Campus*	
14:00	Comida		
15:30	Ponencia Belén Pérez		
16:00	FAQs - Santiago Tomé - David Weinstein - Miguel Carnero - Mª Luz Couce	FAQs - Barry Byrne - Jordi Diaz - Araceli Morales - Eduardo Salido	Taller de Fisioterapia - Virgilia Antón - Alfredo Santalla
18:00 a 20:00	Registro Nacional de Glucogenosis		
18:00 a 21:00	McGym (Gimnasia Glucogenosis Tipo V - McArdle)	**Lugar SAF (Servicio de Actividad Física de la UAB)*	
19:00	Reunión Comité Científico		
19:00	Reunión Asociaciones Glucogenosis Europeas		
21:00	Cena		
	Sábado 7/06		
09:30 a 13:30	Evaluación Médica Glucogenosis Tipo II - Pompe		
10:00	Ponencia David Weinstein		
10:30	Ponencia Antoni L. Andreu		
11:00	Ponencia Gloria Aseguinolaza		
11:30	Pausa Café		
12:00	Ponencia Barry Byrne		
12:30	Ponencia Alberto Molares y Francesc Cayuela		
13:00	Ponencia Tomàs Pinós		
13:30	Ponencia Eurordis		
14:00	Comida		
16:00 16:40	Ponencia David Weinstein	Ponencia Jordi Diaz	Taller "Dudas clínicas" - Tomàs Pinós - Astrid Brull - Noemí de Luna
17:00 17:20	Ponencia Serena Pagliarani Sabrina Lucchiari	Ponencia Virgilia Antón	Ponencia Miguel Carnero
18:00	Clausura V Congreso		
18:30	Asamblea AEEG		
18:00 a 21:00	McGym (Gimnasia Glucogenosis Tipo V - McArdle)	**Lugar SAF (Servicio de Actividad Física de la UAB)*	
21:00	Cena		
	Domingo 8/06		
10:00	Excursión Tibidabo (Barcelona)	**Salida en autocar desde Hotel Campus UAB a las 10:00h, regreso desde Tibidabo a las 18:00h*	

Actividades: Generales | Tipo I / III / IX | Tipo II | Tipo V | Internas | Retransmisión Web vía Streaming | #CongresoAEEG

Hemos contado con una excelente **mesa inaugural** participada por el Excm. Sr. Manel Sabés (Vicerector de Relaciones Institucionales y Territorio UAB) junto con el Sr. Alberto Molares (Presidente de la AEEG) y con el **soporte de la UAB:** Amics UAB, Agencia de Promoción de Actividades i de Congresos, Plataforma de Malalties Minoritàries, Fundació Doctor Robert, Facultad de Traducción e Interpretación, Hotel Campus UAB y del SAF.

En el último año hemos tenido más de **798.000 páginas servidas** y coincidiendo con la celebración del congreso anual el **último mes han sido más de 150.000.**

Estamos especialmente contentos del **éxito de participación** en todos los niveles: 316 inscritos, total de 32 ponentes que han nutrido de contenidos a un programa semanal y compartido experiencias con los asistentes, por primera vez 5 actividades específicas impartidas (Evaluaciones médicas, **Revisiones de visión,** Registro nacional, Rentrenamiento físico y taller de cocina).

Como punto fundamental queremos remarcar los **trabajos y esfuerzos de divulgación** realizados en el marco del congreso: cobertura del acto por RTVE, enviada Nota de Prensa a los medios, Centros de Estudio IES, Hospitales, Centros de Investigación, Asociaciones y entorno UAB.

Ahora empezamos a preparar el **VI Congreso Internacional de Glucogenosis** en Santiago de Compostela, con los objetivos generales de investigación básica, nutrición, fisioterapia y terápias génicas siguiendo con la línea de los dos congresos anteriores, e incorporando la investigación con "Stem Cells" (células madre).